



GALT gene

galactose-1-phosphate uridylyltransferase

Normal Function

The *GALT* gene provides instructions for making an enzyme called galactose-1-phosphate uridylyltransferase. This enzyme enables the body to process a simple sugar called galactose, which is present in small amounts in many foods. Galactose is primarily part of a larger sugar called lactose, which is found in all dairy products and many baby formulas.

Galactose-1-phosphate uridylyltransferase is responsible for one step in a chemical process that breaks down galactose into other molecules that can be used by the body. Specifically, this enzyme converts a modified form of galactose (galactose-1-phosphate) to glucose, which is another simple sugar. Glucose is the main energy source for most cells. This chemical reaction also produces another form of galactose (UDP-galactose) that is used to build galactose-containing proteins and fats. These modified proteins and fats play critical roles in chemical signaling, building cellular structures, transporting molecules, and producing energy.

Health Conditions Related to Genetic Changes

galactosemia

More than 300 mutations in the *GALT* gene have been identified in people with the classic form of galactosemia, a condition that causes life-threatening signs and symptoms beginning shortly after birth. Most of these mutations severely reduce or eliminate the activity of galactose-1-phosphate uridylyltransferase. A shortage of this enzyme prevents cells from processing galactose obtained from the diet. As a result, galactose-1-phosphate and related compounds can build up to toxic levels in the body. The accumulation of these substances damages tissues and organs, leading to the serious medical problems associated with classic galactosemia.

Most changes in the *GALT* gene alter single protein building blocks (amino acids) in galactose-1-phosphate uridylyltransferase. The most common *GALT* mutation in white Europeans and North Americans replaces the amino acid glutamine with the amino acid arginine at position 188 in the enzyme (written as Gln188Arg or Q188R). Another mutation occurs almost exclusively in people of African descent. This genetic change substitutes the amino acid leucine for the amino acid serine at position 135 (written as Ser135Leu or S135L).

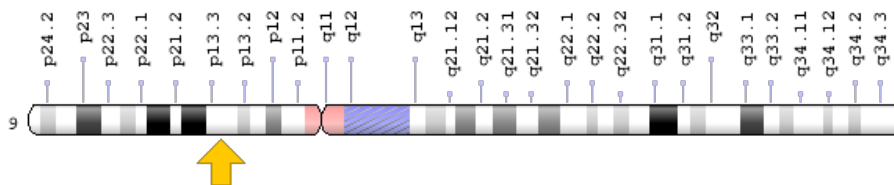
A particular *GALT* mutation called the Duarte variant results in a form of galactosemia with less serious complications than the classic type. This mutation replaces the

amino acid asparagine with the amino acid aspartic acid at protein position 314 (written as Asn314Asp or N314D). The Duarte variant reduces but does not eliminate the activity of galactose-1-phosphate uridylyltransferase. The signs and symptoms associated with this variant tend to be milder because the enzyme retains 5 percent to 20 percent of its normal activity.

Chromosomal Location

Cytogenetic Location: 9p13.3, which is the short (p) arm of chromosome 9 at position 13.3

Molecular Location: base pairs 34,646,589 to 34,650,598 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Gal-1-P uridylyltransferase
- Galactosephosphate Uridylyltransferase
- GALT_HUMAN
- UDP Galactose Pyrophosphorylase
- UTP-Hexose-1-Phosphate Uridylyltransferase
- UTP:alpha-D-hexose-1-phosphate uridylyltransferase

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Figure: UDP-galactose Synthesis and Galactosemia
<https://www.ncbi.nlm.nih.gov/books/NBK1939/figure/ch42.f3/>

GeneReviews

- Classic Galactosemia and Clinical Variant Galactosemia
<https://www.ncbi.nlm.nih.gov/books/NBK1518>
- Duarte Variant Galactosemia
<https://www.ncbi.nlm.nih.gov/books/NBK258640>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GALT%5BTI%5D%29+OR+%28galactose-1-phosphate+uridylyltransferase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- GALACTOSE-1-PHOSPHATE URIDYLYLTRANSFERASE
<http://omim.org/entry/606999>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_GALT.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=GALT%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4135
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2592>
- UniProt
<http://www.uniprot.org/uniprot/P07902>
- University of Utah GALT Mutation Database
http://www.arup.utah.edu/database/GALT/GALT_welcome.php

Sources for This Summary

- Bosch AM, Ijlst L, Oostheim W, Mulders J, Bakker HD, Wijburg FA, Wanders RJ, Waterham HR. Identification of novel mutations in classical galactosemia. *Hum Mutat*. 2005 May;25(5):502. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/15841485>
- Coelho AI, Trabuco M, Ramos R, Silva MJ, Tavares de Almeida I, Leandro P, Rivera I, Vicente JB. Functional and structural impact of the most prevalent missense mutations in classic galactosemia. *Mol Genet Genomic Med*. 2014 Nov;2(6):484-96. doi: 10.1002/mgg3.94. Epub 2014 Jun 23. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/25614870>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4303218/>
- GeneReview: Classic Galactosemia and Clinical Variant Galactosemia
<https://www.ncbi.nlm.nih.gov/books/NBK1518>
- GeneReview: Duarte Variant Galactosemia
<https://www.ncbi.nlm.nih.gov/books/NBK258640>
- McCorvie TJ, Gleason TJ, Fridovich-Keil JL, Timson DJ. Misfolding of galactose 1-phosphate uridylyltransferase can result in type I galactosemia. *Biochim Biophys Acta*. 2013 Aug;1832(8):1279-93. doi: 10.1016/j.bbadis.2013.04.004. Epub 2013 Apr 11. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/23583749>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3679265/>
- McCorvie TJ, Timson DJ. Structural and molecular biology of type I galactosemia: disease-associated mutations. *IUBMB Life*. 2011 Nov;63(11):949-54. doi: 10.1002/iub.510. Epub 2011 Sep 30. Review. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/21960482>
- McCorvie TJ, Timson DJ. The structural and molecular biology of type I galactosemia: Enzymology of galactose 1-phosphate uridylyltransferase. *IUBMB Life*. 2011 Sep;63(9):694-700. doi: 10.1002/iub.511. Epub 2011 Jul 25. Review. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/21793161>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/GALT>

Reviewed: August 2015

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services